



## EDARADD gene

EDAR associated death domain

### Normal Function

The *EDARADD* gene provides instructions for making a protein called the EDAR-associated death domain protein. This protein is part of a signaling pathway that plays an important role in development before birth. Specifically, it is critical for interactions between two embryonic cell layers called the ectoderm and the mesoderm. In the early embryo, these cell layers form the basis for many of the body's organs and tissues. Ectoderm-mesoderm interactions are essential for the formation of several structures that arise from the ectoderm, including the skin, hair, nails, teeth, and sweat glands.

The EDARADD protein interacts with another protein, called the ectodysplasin A receptor, which is produced from the *EDAR* gene. This interaction occurs at a region called the death domain that is present in both proteins. The EDARADD protein acts as an adapter, which means it assists the ectodysplasin A receptor in triggering chemical signals within cells. These signals affect cell activities such as division, growth, and maturation. Before birth, this signaling pathway controls the formation of ectodermal structures such as hair follicles, sweat glands, and teeth.

### Health Conditions Related to Genetic Changes

#### hypohidrotic ectodermal dysplasia

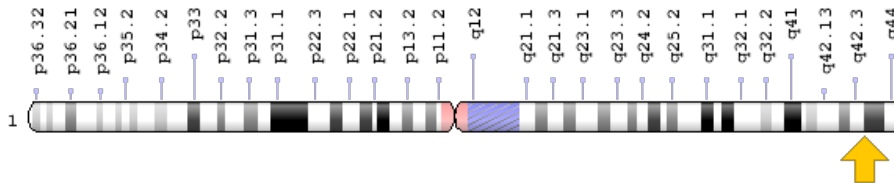
A mutation in the *EDARADD* gene is an infrequent cause of the autosomal recessive form of hypohidrotic ectodermal dysplasia. With autosomal recessive inheritance, two copies of the *EDARADD* gene are mutated in each cell.

The *EDARADD* mutation responsible for hypohidrotic ectodermal dysplasia changes a single protein building block (amino acid) in the EDARADD protein. This genetic change replaces the amino acid glutamine with the amino acid lysine at protein position 142 (written as Glu142Lys). Because this mutation occurs in the death domain, it prevents the EDARADD protein from interacting effectively with the ectodysplasin A receptor. As a result, the receptor probably cannot trigger signals needed for ectoderm-mesoderm interactions and the normal development of hair follicles, sweat glands, and other ectodermal structures. This disruption in ectodermal development leads to the characteristic features of hypohidrotic ectodermal dysplasia.

## Chromosomal Location

Cytogenetic Location: 1q42.3-q43, which is the long (q) arm of chromosome 1 between positions 42.3 and 43

Molecular Location: base pairs 236,394,380 to 236,484,708 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ectodysplasia A receptor associated death domain
- ectodysplasin A receptor associated adapter protein
- EDAD\_HUMAN
- EDAR-associated death domain

## Additional Information & Resources

### GeneReviews

- Hypohidrotic Ectodermal Dysplasia  
<https://www.ncbi.nlm.nih.gov/books/NBK1112>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EDARADD%5BTIAB%5D%29+OR+%28EDAR-associated+death+domain%5BTIAB%5D%29%29+OR+%28%28crinkled+homolog%5BTIAB%5D%29+OR+%28ectodysplasia+A+receptor+associated+death+domain%5BTIAB%5D%29+OR+%28ectodysplasin+A+receptor+associated+adapter+protein%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- EDAR-ASSOCIATED DEATH DOMAIN  
<http://omim.org/entry/606603>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_EDARADD.html](http://atlasgeneticsoncology.org/Genes/GC_EDARADD.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=EDARADD%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=14341](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14341)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/128178>
- UniProt  
<http://www.uniprot.org/uniprot/Q8WWZ3>

## **Sources for This Summary**

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